
DISEASE:
Estrogen resistance syndrome

NAME:	Estrogen resistance syndrome
DESCRIPTION:	Estrogen resistance syndrome is a rare, genetic endocrine disease characterized by estrogen-receptor insensitivity to estrogens and the presence of elevated estrogen and gonadotropin serum levels. Clinical manifestations include absent breast development and primary amenorrhea in association with multicystic ovaries and/or hypoplastic uterus in female patients, normal or abnormal gonadal development in male patients and markedly delayed bone maturation, persistence of open epiphyses, reduced bone mineral density, and variable tall stature in both sexes. Glucose intolerance, hyperinsulinemia and lipid abnormalities may also be present.
ORPHACODE:	785
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>ESR1</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- [Premature Ovarian Failure/Primary Ovarian Insufficiency \(POF/POI\) \(32 genes\)](#)

Related Laboratories

- [Centrum Medische Genetica - UZ Brussel VUB](#)

Related Analytes

- [estrogen receptor 1](#)

Related Gene Panels

- [Premature Ovarian Failure/Insufficiency \(32 genes\) - VUB](#)

Source URL: <http://gentest.healthdata.be/disease/2646>